

## ABSTRACT

### **Detection of genetic morbidity in family practice, using a semi-structured family information interview with a minority population**

*Nadeem Qureshi, University of Nottingham, Queens Medical Centre*

**Objectives:** To evaluate the effectiveness of a culturally sensitive semi-structured interview in family practice to collect family history information from a minority population at increased risk of single gene disorders.

**Subjects and methods:** A case controlled crossover study with pedigree recording by both genetic nurse specialists and a primary care worker. From 1012 records of British Pakistani patients registered with a British Inner City practice, 14 women, identified as having increased genetic risk, were recruited (Group 1). A further 14 age and parity-matched women, with no indication of genetic morbidity in their general practice records (Group 2) were also recruited.

**Results:** Valuable genetic information, not recorded in general practice records, were ascertained through the screening pedigrees; in 4 members of Group 1 (29%) and 6 members of Group 2 (43%). There was poor agreement between the coefficients of consanguinity recorded from pedigrees prepared by the primary care worker and genetic nurse specialists ( $Kappa = 0.157$ ; 95% CI 0.028 – 0.286).

**Conclusion:** Pedigrees can be utilized as a General Practice screening tool to detect relevant genetic morbidity, not ascertained in General Practice records. The process is enhanced when a primary care worker, from the same culture as the volunteers, collects information using the patients' mother tongue. The proactive primary care approach should lead to at-risk couples being able to make informed reproductive choices prior to the birth of their first affected child.